

Dyzostoza kończynowo-czaszkowo-twarzowa

Kod Orpha: 949 Kod OMIM: 201050

Opis choroby *

Definicja

A very rare acrofacialdyosotosis characterized by short stature, acrocephaly, ocular hypertelorism, ptosis of eyelids, ocular proptosis, downslanting palpebral fissures, high nasal bridge, anteverted nostrils, short philtrum, cleft palate, micrognathia, abnormal external ears, preauricular pits, mixed hearing loss, bulbous digits, metatarsus varus, pectus excavatum and various radiological abnormalities. Features of this syndrome were reported to overlap with otopalatodigital syndrome types 1 and 2. There have been no further descriptions in the literature since 1988.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Kaplan-Plauchu-Fitch syndrome
Zespół Kaplana, plauchu i Fitcha

Kod ORPHA

949

Kod OMIM

201050

Kod ICD10

Q87.0

Kod ICD11

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[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

Dostępna na stronie www.orphanet.pl